

GEPHE SUMMARY

	Gephebase Gene		GepheID
agrecan (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~agrecan~#gephebase-summary-title)		GP00000072	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category			
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)			Trait	
Body size (height) (https://www.gephebase.org/search-criteria?/and+Trait=~Body size (height)~#gephebase-summary-title)				
	Trait State in Taxon A			
Homo sapiens			Trait State in Taxon B	
Homo sapiens				
	Ancestral State			
Data not curated			Taxonomic Status	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific~#gephebase-summary-title)				
	Taxon A		Taxon B	
	Latin Name			Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens~#gephebase-summary-title)			Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens~#gephebase-summary-title)	
	Common Name			Common Name
human			human	
	Synonyms			Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens			human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens	
	Rank			Rank
species			species	
	Lineage			Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo			cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo	
	Parent			Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)			Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	
	NCBI Taxonomy ID			NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)			9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	
	is Taxon A an Intraspecies?			is Taxon B an Intraspecies?
No			No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
ACAN		P16112 (http://www.uniprot.org/uniprot/P16112)	
	Synonyms		GenebankID or UniProtKB
AGC1; SEDK; AGCAN; CSPG1; MSK16; CSPGCP; SSOAOD		M55172 (https://www.ncbi.nlm.nih.gov/nuccore/M55172)	
	String		
9606.ENSPO0000387356 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000387356)			
	Sequence Similarities		
Belongs to the aggrecan/versican proteoglycan family.			
	GO - Molecular Function		
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)			
GO:0030246 : carbohydrate binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030246)			
GO:0005201 : extracellular matrix structural constituent (https://www.ebi.ac.uk/QuickGO/term/GO:0005201)			

GO:0030021 : extracellular matrix structural constituent conferring compression resistance
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030021>)
GO:0005540 : hyaluronic acid binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005540>)

GO - Biological Process

GO:0007155 : cell adhesion (<https://www.ebi.ac.uk/QuickGO/term/GO:0007155>)
GO:0007417 : central nervous system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007417>)
GO:0001502 : cartilage condensation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001502>)
GO:0002063 : chondrocyte development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002063>)
GO:0030199 : collagen fibril organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030199>)
GO:0030198 : extracellular matrix organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030198>)
GO:0007507 : heart development (<https://www.ebi.ac.uk/QuickGO/term/GO:0007507>)
GO:0018146 : keratan sulfate biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0018146>)
GO:0042340 : keratan sulfate catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042340>)
GO:0030166 : proteoglycan biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030166>)
GO:0006508 : proteolysis (<https://www.ebi.ac.uk/QuickGO/term/GO:0006508>)
GO:0001501 : skeletal system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001501>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0062023 : collagen-containing extracellular matrix
(<https://www.ebi.ac.uk/QuickGO/term/GO:0062023>)
GO:0005604 : basement membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005604>)
GO:0031012 : extracellular matrix (<https://www.ebi.ac.uk/QuickGO/term/GO:0031012>)
GO:0098982 : GABA-ergic synapse (<https://www.ebi.ac.uk/QuickGO/term/GO:0098982>)
GO:0098978 : glutamatergic synapse
(<https://www.ebi.ac.uk/QuickGO/term/GO:0098978>)
GO:0005796 : Golgi lumen (<https://www.ebi.ac.uk/QuickGO/term/GO:0005796>)
GO:0043202 : lysosomal lumen (<https://www.ebi.ac.uk/QuickGO/term/GO:0043202>)
GO:0098966 : perisynaptic extracellular matrix
(<https://www.ebi.ac.uk/QuickGO/term/GO:0098966>)

	Presumptive Null
Unknown (#gephebase-summary-title)	
	Molecular Type
Unknown (#gephebase-summary-title)	
	Aberration Type
Unknown (#gephebase-summary-title)	
	Molecular Details of the Mutation
unknown	
	Experimental Evidence
Association Mapping (#gephebase-summary-title)	
	Main Reference
Recent progress in the study of the genetics of height. (2011) (https://pubmed.ncbi.nlm.nih.gov/21340692)	
	Authors
Lettre G	
	Abstract
Adult height is a classic polygenic trait of high narrow-sense heritability ($h^2 = 0.8$). In the late nineteenth to early twentieth century, variation in adult height was used as a model to set the foundation of the fields of statistics and quantitative genetics. More recently, with our increasing knowledge concerning the extent of genetic variation in the human genome, human geneticists have used genome-wide association studies to identify hundreds of loci robustly associated with adult height, providing new insights into human growth and development, and into the architecture of complex human traits. In this review, I highlight the progress made in the last 2 years in understanding how genetic variation controls height variation in humans, including non-Caucasian populations and children.	
	Additional References

RELATED GEPHE

	Related Genes
23 (ADAMTS10, CREBRF, DC-STAMP domain containing 2 (DCST2), DYM, EIF2AK3, FTO, GDF5, GHSR, GPR133, Growth Hormone Receptor (GHR), HMGA2, Insulin-like growth factor receptor 1 (IGF1R), JAZF1, KCNQ1, LCOLR, LIN28B, natriuretic peptide precursor type C (NPPC), natriuretic peptide receptor 3 (NPR3), Patched1 (Ptc1), PPAR-delta, TRIP11 (=GMAP-210), SMAD family member 2 (SMAD2), stanniocalcin 2 (STC2)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~9606#/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title)	
	Related Haplotypes
No matches found.	

EXTERNAL LINKS

